Alaska Birth Defect Prevalence Estimates (per 10,000 live births), 2008-2012 birth years (N = 56,309)

Nationally reportable major congenital anomalies (N=47)

Defect	Reports	Report Prevalence (95%CI)	PPV	NPV	Estimated Defects	Defect Prevalence (95%CI)*
Anencephalus	12	2.1 (1.1,3.7)	0.3929	1.0000	4.7	0.8 (0.3,1.8)
Anophthalmia / microphthalmia	9	1.6 (0.7,3.0)				NA
Anotia / microtia	23	4.1 (2.6,6.1)				NA
Aortic valve stenosis	11	2.0 (1.0,3.5)				NA
Atrial septal defect	902	160.2 (149.9,171.0)				NA
Atrioventricular septal defect	36	6.4 (4.5,8.9)				NA
Biliary atresia	14	2.5 (1.4,4.2)				NA
Bladder exstrophy	-	-				NA
Choanal atresia	18	3.2 (1.9,5.1)				NA
Cleft lip alone	22	3.9 (2.4,5.9)				NA
Cleft lip with celft palate	49	8.7 (6.4,11.5)				NA
Cleft palate alone	74	13.1 (10.3,16.5)				NA
Cloacal exstrophy	-	-				NA
Clubfoot	208	36.9 (32.1,42.3)				NA
Coarctation of the aorta	25	4.4 (2.9,6.6)				NA
Common truncus	16	2.8 (1.6,4.6)				NA
Congenital cataract	31	5.5 (3.7,7.8)				NA
Congenital posterior urethral valves	49	8.7 (6.4,11.5)				NA
Craniosynostosis	-	-				NA
Deletion 22 q11	-	-				NA
Diaphragmatic hernia	31	5.5 (3.7,7.8)				NA
Double outlet right ventricle	10	1.8 (0.9,3.3)				NA
Ebstein anomaly	9	1.6 (0.7,3.0)				NA
Encephalocele	16	2.8 (1.6,4.6)	0.3778	1.0000	6.4	1.1 (0.5,2.3)
Esophageal atresia / tracheoesophageal fistula	15	2.7 (1.5,4.4)				NA
Gastroschisis	29	5.2 (3.4,7.4)	0.6174	1.0000	20.5	3.6 (2.3,5.5)
Holoprosencephaly	57	10.1 (7.7,13.1)				NA
Hypoplastic left heart syndrome	7	1.2 (0.5,2.6)				NA
Hypospadias	330	113.6 (101.7,126.5)	0.8618	0.9996	294.5	101.4 (90.3,113.4)
Inerrupted aortic arch	25	4.4 (2.9,6.6)				NA
Limb deficiencies	66	11.7 (9.1,14.9)				NA
Omphalocele	70	12.4 (9.7,15.7)	0.1856	1.0000	13.1	2.3 (1.4,3.9)

Defect	Reports	Report Prevalence (95%CI)	PPV	NPV	Estimated Defects	Defect Prevalence (95%CI)*
Pulmonary valve atresia and stenosis	79	14.0 (11.1,17.5)				NA
Rectal and large intestinal atresia / stenosis	56	9.9 (7.5,12.9)				NA
Renal agenesis / hypoplasia	51	9.1 (6.7,11.9)				NA
Single ventricle	5	0.9 (0.3,2.1)				NA
Small intestinal atresia / stenosis	37	6.6 (4.6,9.1)				NA
Spina bifida without anencephalus	15	2.7 (1.5,4.4)	0.5946	0.9999	12.4	2.2 (1.2,3.7)
Teratology of fallot	30	5.3 (3.6,7.6)				NA
Total anomalous pulmonary venous sonnection	10	1.8 (0.9,3.3)				NA
Transposition of the great arteries	21	3.7 (2.3,5.7)				NA
Tricuspid valve atresia and stenosis	-	-				NA
Trisomy 13	9	1.6 (0.7,3.0)	0.5000	1.0000	4.5	0.8 (0.3,1.8)
Trisomy 18	12	2.1 (1.1,3.7)	0.5833	1.0000	7.0	1.2 (0.5,2.6)
Trisomy 21 (Down syndrome)	100	17.8 (14.4,21.6)	0.8182	0.9999	87.3	15.5 (12.5,19.1)
Turner syndrome	48	8.5 (6.3,11.3)	0.5217	1.0000	26.8	4.8 (3.2,6.8)
Ventricular septal defect	570	101.2 (93.1,109.9)				NA

Ref: NA = Information not available as of publication; '-'= supressed for cell counts <5; PPV = Positive Predicitve Value; NPV = Negative Predictive value; 95% CI = 95% Confidence Interval

Note: The Alaska Birth Defects Registry (ABDR) was established and operates under Alaska statute 7 AAC 27.012. Statue allows reports to by age six years of a child. This report includes defects reported before the 3rd birthday.

Column descriptions:
Defect = The Nationally reportable birth defect grouping name.
Reports = Unless otherwise noted, the number of unique reports by child recieved by ABDR during the specified birth years.
Report Prevalence (95% CI) = The number of reported defects divided by the number of Alaska resident in-state births (Children can be reported multiple times. they are counted only once for each condition).
PPV = The probability of being defect positive given being reported for the defect.
NPV = The probability of being defect negative given not being reported for the defect.
Estimated Defects = The estimated number of defects based on the report prevalence, PPV, and 1-NPV.
Defect Prevalence (95% CI) = The estimated defect prevalence calculated using a Bayesian approach based on the reported prevalence, PPV and 1-NPV (see formula below).

*Through medical records review and case confirmation of a random sample of reported cases, the defect prevalence is calculated as:

$$PPV(PositivePredictiveValue) = p(defect)|report)$$
$$NPV(NegativePredictiveValue) = p(\overline{defect}|\overline{report})]$$

$$p(defect) \approx [p(report) \cdot PPV] + [p(\overline{report}) \cdot 1 - NPV)]$$

Defect prevalence estimates are a more accurate estimation of the actual diagnosed prevelance of birth defects compared to the reported prevelance estimates in Alaska. ABDR obtains reports from medical providers using International Classification of Disease (ICD) codes that are extracted from individual systems which when aggregated may not reflect true diagnostics. Caution should be used when interpreting and comparing the reported prevalence estimates with national estimates.

Condition Notes:
Cleft Lip (CL), Cleft Palate (CP), and Cleft Lip and Palate (CLP) are coded to be mutually exclusive groups.Because an individual can be reported multiple times and for any condition, this coding ensures that for these cleft conditions the classifications conform to the expectation of being mutually exclusive.

- Cleft lip alone includes reports without Cleft palate (CP) or cleft lip and palate (CLP)
- Cleft palate alone includes reports without Cleft lip (CL) or cleft lip and palate (CLP)
- Cleft lip and palate includes reports of both Cleft lip (CL) and Cleft palate (CP), Cleft lip and palate (CLP) or any other combination of Cleft lip (CL) or Cleft palate (CP) with cleft lip and palate (CLP)

Hypospadias is restricted to male births (N = 29,054).

Resources:
National Birth Defects Prevention Network
Centers for Diesase Control and Prevention

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Code source: `R:\ABDR\Analysis_New\ABDR_CASECONF\Prevalence08.12.Rmd`